

Chapter 15: The Chromosomal Basis of Inheritance

Concept 15.1 Mendelian inheritance has its physical basis in the behavior of chromosomes

1. What is the *chromosome theory of inheritance*?

The chromosome theory of inheritance is a basic principle in biology stating that genes are located at specific positions (loci) on chromosomes and that the behavior of chromosomes during meiosis accounts for inheritance patterns.

2. Explain the *law of segregation*. Use two different colored pencils to illustrate the segregation of alleles. You may want to consult Figure 15.2 in your text, and model your sketches on this.

See page 287 in your text for the labeled figure.

The law of segregation states that the two alleles for each gene separate during gamete formation.

3. Explain the *law of independent assortment*. To demonstrate that you understand this concept, consider a cell with two pairs of chromosomes. Sketch the two different ways these chromosomes might be arranged during metaphase I.

See page 287 in your text for the labeled figure.

The law of independent assortment states that each pair of alleles segregates, or assort, independently of each other pair during gamete formation; applies when genes for two characters are located on different pairs of homologous chromosomes or when they are far enough apart on the same chromosome to behave as though they are on different chromosomes.

4. *Thomas Hunt Morgan* selected *Drosophila melanogaster* as his experimental organism. List at least three reasons the fruit fly is an excellent subject for genetic studies.

1. Single mating will produce hundreds of offspring.
2. A new generation can be bred every two weeks.
3. The fruit fly has only four pairs of chromosomes, which are easily distinguishable with a light microscope.

5. The notation for *wild type* and *mutant* traits follows some accepted conventions. Notate the following genotypes for a female fruit fly:

a. a fly homozygous for red eyes $X^{w+}X^{w+}$

b. a fly heterozygous for red eyes $X^{w+}X^w$

c. a fly homozygous for white eyes X^wX^w

6. When Thomas Hunt Morgan mated a white-eyed male fly with a red-eyed female, he came to the startling conclusion that the trait for eye color was located on the chromosome that determines sex. Show this cross. Begin with the parental generation, and go through the F₂.

See page 289 of your text for the labeled figure.

parental generation: A red-eyed female was crossed with a white-eyed male.

F₁ generation: All the offspring of the P generation had red eyes

F₂ generation: The offspring showed a ratio of three red-eyed flies to one white-eyed fly. However, there were no white-eyed females.

7. What unusual result suggested that the eye-color trait is located on the X chromosome?

The white-eye trait showed up only in males.

Concept 15.2 Sex-linked genes exhibit unique patterns of inheritance

8. There are several variations in the way sex is determined in different species. Complete the following figure to explain four different methods of sex determination.

See page 290 of your text for the labeled figure.

9. What is the *SRY* gene? Where is it found, and what does it do?

SRY refers to the Sex determining Region of Y, a gene found on the Y chromosome that is required for the development of testes.

10. What is the definition of a *sex-linked gene*?

A gene located on either sex chromosome. Most sex-linked genes are on the X chromosome and show distinctive patterns of inheritance; there are very few genes on the Y chromosome.

11. In humans, how has that term been historically modified?

Typically, the sex-linked traits are actually X-linked genes.

12. Name and describe three human sex-linked disorders.

1. Duchenne muscular dystrophy: A human genetic disease caused by a sex-linked recessive allele; characterized by progressive weakening and a loss of muscle tissue
2. Hemophilia: A human genetic disease caused by a sex-linked recessive allele, resulting in the absence of one or more blood-clotting proteins; characterized by excessive bleeding following injury
3. Color Blindness: A mild disorder almost always inherited as an X-linked allele

13. Try the following problem (Figure 15.7b in your text). A female who carries an allele for color blindness, but who is not color-blind, mates with a male who has normal color vision. What is the probability that they will have a son who is color-blind? A Punnett square to use for this problem is shown in the following figure.

See page 291 of your text for the labeled figure.

If a carrier mates with a male who has normal color vision, there is a 50% chance that each daughter will be a carrier like her mother and a 50% chance that each son will have the disorder.

14. What is a *Barr body*? Why do human females show a Barr body in their cells?

A Barr body is a dense object lying along the inside of the nuclear envelope in cells of female mammals, representing a highly condensed, inactivated X chromosome. Female mammals, including humans, inherit two X chromosomes—twice the number inherited by males; females show a Barr body in their cells so that the cells of females and males have the same effective dose (one copy) of most X-linked genes.

15. X inactivation maintains the proper gene dosage. How is the X chromosome inactivated?

The selection of which X chromosome will form the Barr body occurs randomly and independently in each embryonic cell present at the time of X inactivation.

16. Why can you say that all calico cats are females?

The tortoiseshell gene is on the X chromosome, and the tortoiseshell phenotype requires the presence of two different alleles, one for orange fur and one for black fur. Normally, only females can have both alleles, because only they have two X chromosomes.

Concept 15.3 Linked genes tend to be inherited together because they are located near each other on the same chromosome

17. What are *linked genes*? Do linked genes sort independently?

Linked genes are genes located close enough together on a chromosome that they tend to be inherited together. These genes do not sort independently, but rather are transmitted as a unit. It is important to note that as Morgan's experiments illustrated, some mechanism (later discovered to be "crossing over") occasionally breaks the linkage between specific alleles of genes on the same chromosome.

18. If two genes are linked on the same chromosome, we call this combination the *parental combination*. These genes will be transmitted as a unit and will not sort independently. However, during meiosis, *crossing over* occurs between homologous chromosomes, and the linked genes can become "unlinked." In general, the farther two genes are from each other along the chromosome, the more often they will come "unlinked." *Genetic recombination* is the process during which linked genes become unlinked. What do geneticists call the offspring that show these new combinations?

Geneticists call the offspring that show these new combinations recombinant types, or recombinants for short.

19. Review meiosis. When does *crossing over* occur? **Prophase I**
20. *Alfred H. Sturtevant*, a student of Thomas Hunt Morgan, used assumptions from observations of crossovers to map genes. What is a *linkage map*?

A linkage map is a genetic map based on the frequencies of recombination between markers during crossing over of homologous chromosomes.

21. What is a *map unit*?

A map unit is a unit of measurement of the distance between genes. One map unit is equivalent to a 1% recombination frequency.

22. Use the figure below, which is from Figure 15.10. It shows the results of a cross between a fruit fly that is heterozygous for a gray body with normal wings, and a fruit fly that has a black body with vestigial wings. Because these genes are linked, the results are not what might have been predicted. Show the phenotypes and number of each type of offspring. Indicate which offspring are the recombinants and which are the parental type. Finally, calculate the map distance between the two genes. Show all your work here.

See page 295 in your text for the labeled figure.

Since 391 of the total 2,300 offspring do not show the parental phenotypes, they are recombinants. Since the frequency of recombination is therefore 17%, the two genes are 17 map units apart.

Concept 15.4 Alterations of chromosome number or structure cause some genetic disorders

23. What occurs in *nondisjunction*?

Nondisjunction is an error in meiosis or mitosis in which members of a pair of homologous chromosomes or a pair of sister chromatids fail to separate properly from each other.

24. Explain each of the following terms:

aneuploidy: A chromosomal aberration in which one or more chromosomes are present in extra copies or are deficient in number. Trisomy 21 (Down syndrome) is an aneuploidy.

monosomy: Refers to a diploid cell that has only one copy of a particular chromosome instead of the normal two. Turner syndrome is a human monosomy; the female has only one X chromosome.

trisomy: Refers to a diploid cell that has three copies of a particular chromosome instead of the normal two. Trisomy 21 is trisomic for chromosome 21.

polyploidy: A chromosomal alteration in which the organism possesses more than two complete chromosome sets. It is the result of an accident of cell division.

25. What causes Down *syndrome*? What are four characteristics of Down syndrome?

Down syndrome is usually the result of an extra chromosome 21, so that each cell has a total of 47 chromosomes. Because the cells are trisomic for chromosome 21, Down syndrome is often called trisomy 21.

Four characteristics of Down syndrome:

1. Characteristic facial features
2. Short stature
3. Correctable heart defects
4. Developmental delays

26. For each of the following human aneuploidies, give the sex of the individual as well as any physical manifestation of the syndrome.

	Sex	Physical Traits
XXY	Male	Klinefelter syndrome: male sex organs, but abnormally small/ sterile testes; some breast enlargement and other female characteristics
XXX	Female	Trisomy X: no unusual physical features other than being slightly taller than average; at risk for learning disabilities; fertile
XO	Female	Turner syndrome: phenotypically female, but sterile due to lack of maturation in sex organs; secondary sex characteristics developed with estrogen replacement; normal intelligence
XYY	Male	Normal sexual development; taller than average stature

27. Chromosome structure can be altered in several ways. Label each type of alteration shown in this figure, and explain what occurs.

See page 298 in your text for the labeled figure.

deletion: A deficiency in a chromosome resulting from the loss of a fragment through breakage

duplication: An aberration in chromosome structure due to fusion with a fragment from a homologous chromosome, such that a portion of a chromosome is duplicated

inversion: An aberration in chromosome structure resulting from reattachment of a chromosomal fragment in a reverse orientation to the chromosome from which it originated

translocation: An aberration in chromosome structure resulting from attachment of a chromosomal fragment to a nonhomologous chromosome

Concept 15.5 Some inheritance patterns are exceptions to the standard Mendelian inheritance

28. A number of genes will cause a variation in phenotype, depending on whether the gene came from the father or the mother. This variation occurs because of *genomic imprinting*. Explain genomic imprinting.

Genomic imprinting occurs during gamete formation and results in the silencing of a particular allele of certain genes. Because these genes are imprinted differently in sperm and eggs, the zygote expresses only one allele of an imprinted gene that is inherited from either the female or the male parent. The imprints are then transmitted to all body cells during development.

29. Although you inherited one chromosome of each pair from your mother and your father, you have inherited a group of genes from your mother only. What genes are these?

Mitochondrial DNA

30. You should have identified mitochondrial DNA as the correct response to question 29 above. What other organelle has its own genes? These are *extranuclear genes*.

Chloroplasts or mitochondria

Test Your Understanding Answers

Just as in Chapter 14, this section asks you to do a series of problems. One way to determine your understanding of genetics is to work many genetics problems, so don't skip this important step! Complete the following questions from the problems at the end of the chapter: Questions 1-4, 7-13.

Work neatly, and show all work. As you know, you can check your solutions in your text.

See page A-14 (Appendix) for answers.